**Current Project Title:** Disease Severity and Its Effect on Health-Quality of Life and Meaning-Making in Families Affected by Smith Lemli Opitz

**Purpose:** To understand the lived experience of Smith-Lemli Opitz through the lens of health-related quality of life and how it relates to the silver linings and challenges of patients and their families.

* To study the potential correlation between health-quality of life and disease severity in SLOS.
* To learn more about the psychosocial impacts SLOS has on patients and their families.
* To find the meanings/lessons having a child with SLOS has brought to families.

**What are the (main) research questions are you trying to answer? List them.**

* How does disease severity and its effect on quality of life reflect on parents’ ability to find meaning in their child’s Smith Lemli Opitz diagnosis?
* Does disease severity affect quality of life?
* Do other factors like income, race, and location affect parents’ perceptions of their children?

Disease Severity and Meaning Making in SLOS

Start of Block: Introduction

My name is Riley Schweig, and I am a genetic counseling student at Boston University School of Medicine. I invite you to participate in a research study (H-41650) exploring the lived experience of Smith-Lemli Opitz through the lens of health-related quality of life and how it relates to the silver linings and challenges of patients and their families. The data will be analyzed to find the potential correlation between health-quality of life and disease severity in SLOS, to learn more about the psychosocial impacts SLOS has on patients and their families, and to potentially find any meanings or lessons having a child with SLOS has brought to families.   
  
  
Any English-speaking person who is 18 years or older with at least one child affected by SLOS is invited to participate. If you have more than one child affected, we ask that you only reflect on your oldest child with SLOS.   
  
  
 The online survey is anonymous and will take 10-15 minutes to complete. Participation is voluntary and you may choose to exit the survey at any point if you wish to no longer participate. All your responses will remain anonymous and there is no way to track your responses back to you. In appreciation for your time and effort, a donation will be made to the SLOS foundation.   
  
  
This research study was reviewed by the Boston University Medical Campus Institutional Review Board. If you have any questions or concerns, please contact the IRB at: medirb@bu.edu or email me at rschweig@bu.edu. Thank you for your contribution!

End of Block: Introduction

Start of Block: Demographics

Q1 How many children do you care for in total?

* 1 (1)
* 2 (2)
* 3 (3)
* 4 (4)
* 5+ (5)

Display This Question:

If How many children do you care for in total? = 1

Q2 Please indicate your relationship to your child

* Mother (1)
* Father (2)
* Sibling (3)
* Grandparent (4)
* Other (5)

Display This Question:

If How many children do you care for in total? = 5+

Q3 Please indicate your relationship to your children

* Mother (1)
* Father (2)
* Sibling (3)
* Grandparent (4)
* Other (5)

Display This Question:

If How many children do you care for in total? = 2

Q4 Please indicate your relationship to the children you care for

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
|  | Mother (1) | Father (2) | Sibling (3) | Grandparent (4) | Other (5) |
| Eldest Child (1) |  |  |  |  |  |
| Child 2 (2) |  |  |  |  |  |

Display This Question:

If How many children do you care for in total? = 3

Q5 Please indicate your relationship to the children you care for

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
|  | Mother (1) | Father (2) | Sibling (3) | Grandparent (4) | Other (5) |
| Eldest Child (1) |  |  |  |  |  |
| Child 2 (2) |  |  |  |  |  |
| Child 3 (5) |  |  |  |  |  |

Display This Question:

If How many children do you care for in total? = 4

Q6 Please indicate your relationship to the children you care for

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
|  | Mother (1) | Father (2) | Sibling (3) | Grandparent (4) | Other (5) |
| Eldest Child (1) |  |  |  |  |  |
| Child 2 (2) |  |  |  |  |  |
| Child 3 (5) |  |  |  |  |  |
| Youngest child (6) |  |  |  |  |  |

Q7 How many of the children you care for have Smith Lemli Opitz syndrome (SLOS)?

* 1 (1)
* 2 (2)
* 3 (3)
* 4 (4)
* 5+ (5)

Display This Question:

If How many of the children you care have Smith Lemli Opitz syndrome (SLOS)? = 1

Q8 How old is your child with SLOS?

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Display This Question:

If How many of the children you care have Smith Lemli Opitz syndrome (SLOS)? = 2

Or How many of the children you care have Smith Lemli Opitz syndrome (SLOS)? = 3

Or How many of the children you care have Smith Lemli Opitz syndrome (SLOS)? = 4

Or How many of the children you care have Smith Lemli Opitz syndrome (SLOS)? = 5+

Q9 How old is your eldest child with SLOS?

\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

Display This Question:

If How many of the children you care have Smith Lemli Opitz syndrome (SLOS)? = 2

Or How many of the children you care have Smith Lemli Opitz syndrome (SLOS)? = 3

Or How many of the children you care have Smith Lemli Opitz syndrome (SLOS)? = 4

Or How many of the children you care have Smith Lemli Opitz syndrome (SLOS)? = 5+

Q10 How old is your youngest child with SLOS?

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Q11 Please indicate what ethnicity you most identify with

* White (1)
* Hispanic or Latino (2)
* Black or African American (3)
* Native American (4)
* Asian or Pacific Islander (5)
* Other (6)
* Care not to respond (7)

Q12 How would you describe the area where your family lives?

* Urban (1)
* Suburban (2)
* Rural (3)

Q13 Please indicate the answer that includes your entire household income in (previous year) before taxes.

* Less than $10,000 (1)
* $10,000 to $19,999 (2)
* $20,000 to $29,999 (3)
* $30,000 to $39,999 (4)
* $40,000 to $49,999 (5)
* $50,000 to $59,999 (6)
* $60,000 to $69,999 (7)
* $70,000 to $79,999 (8)
* $80,000 to $89,999 (9)
* $90,000 to $99,999 (10)
* $100,000 to $149,999 (11)
* $150,000 or more (12)
* Prefer not to answer (13)

End of Block: Demographics

Start of Block: Block 6

Display This Question:

If How many of the children you care have Smith Lemli Opitz syndrome (SLOS)? = 2

Or How many of the children you care have Smith Lemli Opitz syndrome (SLOS)? = 3

Or How many of the children you care have Smith Lemli Opitz syndrome (SLOS)? = 4

Or How many of the children you care have Smith Lemli Opitz syndrome (SLOS)? = 5+

Q14 Please only fill out the remainder of the survey for your oldest child that is affected by SLOS.

End of Block: Block 6

Start of Block: Disease Severity

Q15 How old was your child when they were diagnosed with SLOS?

* Prenatally (1)
* Before 1 years old (2)
* 1-5 years old (3)
* 6+ (4)

Q16 Please select all organ systems below that your child had affected at the time of diagnosis.

* Brain (1)
* Mouth (2)
* Feet/toes (3)
* Eyes (4)
* Heart (5)
* Kidney (6)
* Liver (7)
* Lungs (8)
* Bowels (9)
* Genitalia (10)
* I do not know (11)

Display This Question:

If Please select all organ systems below that your child had affected at the time of diagnosis. = Brain

Q17 Please select what differences your child had with their brain at time of diagnosis.

* Seizures (1)
* Structural defects (2)
* None of the above (3)

Display This Question:

If Please select all organ systems below that your child had affected at the time of diagnosis. = Mouth

Q18 Please select what differences your child had with their mouth at time of diagnosis.

* Bifid uvula (1)
* Cleft lip and/or cleft palate (2)
* None of the above (3)

Display This Question:

If Please select all organ systems below that your child had affected at the time of diagnosis. = Feet/toes

Q19 Please select what differences your child had with their feet/toes at time of diagnosis.

* 2/3 toe syndactyly (1)
* Club foot (2)
* None of the above (3)

Display This Question:

If Please select all organ systems below that your child had affected at the time of diagnosis. = Eyes

Q20 Does your child have cataracts?

* Yes (1)
* No (2)

Display This Question:

If Please select all organ systems below that your child had affected at the time of diagnosis. = Kidney

Q21 Please select what differences your child had with their kidney(s) at time of diagnosis.

* Kidney disease (1)
* Kidney agenesis (2)
* None of the above (3)

Display This Question:

If Please select all organ systems below that your child had affected at the time of diagnosis. = Liver

Q22 Please select what differences your child had with their liver at time of diagnosis.

* Structural abnormality (1)
* Progressive liver disease (2)
* None of the above (3)

Display This Question:

If Please select all organ systems below that your child had affected at the time of diagnosis. = Lungs

Q23 Please select what differences your child had with their lungs at time of diagnosis.

* Pulmonary hypoplasia (1)
* Pulmonary cysts (2)
* None of the above (3)

Display This Question:

If Please select all organ systems below that your child had affected at the time of diagnosis. = Bowels

Q24 Please select what differences your child had with their bowels at time of diagnosis.

* Pyloric stenosis (1)
* Hischsprung disease (2)
* None of the above (3)

Display This Question:

If Please select all organ systems below that your child had affected at the time of diagnosis. = Genitalia

Q25 Please select what differences your child had with their genitalia at time of diagnosis.

* Hypospadias (1)
* Ambiguous genitalia (2)
* None of the above (3)

Q26 Please select all behavioral features your child has from the list below:

* Low muscle tone (1)
* Inconsistent sleeping pattern (2)
* Screams for a long time (3)
* Aggressive outbursts/temper tantrums (4)
* Need for extra academic support in school (5)
* Language delays (6)
* None of the above (7)

Display This Question:

If Please select all behavioral features your child has from the list below: = Need for extra academic support in school

Q27 Please select what extra help your child receives in school

* IEP (1)
* Applied Behavioral Therapy (ABA) (2)
* Occupational therapy (3)
* Physical therapy (4)
* Speech therapy (5)
* No extra help is provided (6)
* Other (7) \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

End of Block: Disease Severity

Start of Block: PedsQL inventory

Q28 In the past one month, how often of a problem has your child had with...

|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
|  | Never (1) | Almost Never (2) | Sometimes (3) | Often (4) | Almost Always (5) | N/A (6) |
| Walking more than one block (1) |  |  |  |  |  |  |
| Running (2) |  |  |  |  |  |  |
| Participating in sports or exercise (3) |  |  |  |  |  |  |
| Lifting something heavy (4) |  |  |  |  |  |  |
| Taking a bath or shower by him or herself (5) |  |  |  |  |  |  |
| Doing chores (6) |  |  |  |  |  |  |
| Having hurts or aches (7) |  |  |  |  |  |  |
| Low energy level (8) |  |  |  |  |  |  |

Q29 In the past one month, how often of a problem has your child had with...

|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
|  | Never (1) | Almost Never (2) | Sometimes (3) | Often (4) | Almost Always (5) | N/A (6) |
| Feeling afraid or scared (1) |  |  |  |  |  |  |
| Feeling sad or blue (2) |  |  |  |  |  |  |
| Feeing angry (3) |  |  |  |  |  |  |
| Trouble sleeping (4) |  |  |  |  |  |  |
| Worrying about what will happen to them (5) |  |  |  |  |  |  |

Q30 In the past one month, how often of a problem has your child had with...

|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
|  | Never (1) | Almost Never (2) | Sometimes (3) | Often (4) | Almost Always (5) | N/A (6) |
| Getting along with other children (1) |  |  |  |  |  |  |
| Other kids not wanting to be their friend (2) |  |  |  |  |  |  |
| Getting teased by other children (3) |  |  |  |  |  |  |
| Not able to do things that other children their age can do (4) |  |  |  |  |  |  |
| Keeping up when playing with other children (5) |  |  |  |  |  |  |

Q31 In the past one month, how often of a problem has your child had with...

|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
|  | Never (1) | Almost Never (2) | Sometimes (3) | Often (4) | Almost Always (5) | N/A (6) |
| Paying attention in class (1) |  |  |  |  |  |  |
| Forgetting things (2) |  |  |  |  |  |  |
| Keeping up with school activities (3) |  |  |  |  |  |  |
| Missing school because of not feeling well (4) |  |  |  |  |  |  |
| Missing school to go to doctor or hospital (5) |  |  |  |  |  |  |

End of Block: PedsQL inventory

Start of Block: Challenges

Q32 Please select any of the following challenges your family has experienced due to SLOS:

* Mental strain (Ex: Stress, worry, anger, frustration, etc.) (1)
* Physical strain (Ex: Exhaustion, headaches, etc.) (2)
* Relationship strain (Ex:Difficulties with your partner, other children, other family members, etc.) (3)
* Financial strain (Ex:Reduced work hours, large medical bills, increased doctors visits, etc.) (5)
* Other (4) \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_
* None of the above (6)

End of Block: Challenges

Start of Block: Meaning-Making

Q33 If you could give three words to describe your child, what would they be?

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Q34 What positive impacts has having a child with SLOS had on you and your family?

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Q35 What has having a child with SLOS taught you and your family?

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End of Block: Meaning-Making